EGR2 gene

early growth response 2

Normal Function

The *EGR2* gene provides instructions for making a protein called early growth response 2, which is part of the early growth response family of proteins. These proteins bind to specific areas of DNA and help control the activity of particular genes. On the basis of this action, the proteins are called transcription factors.

The early growth response 2 protein activates several genes that are involved in the formation and maintenance of myelin, the protective substance that covers nerve cells. Myelin is essential for the efficient transmission of nerve impulses.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

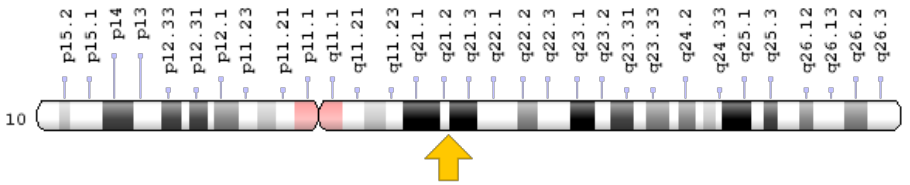
Mutations in the *EGR2* gene can cause two forms of Charcot-Marie-Tooth disease, type 1D or type 4E (sometimes called congenital hypomyelinating neuropathy). These mutations change single protein building blocks (amino acids) in the early growth response 2 protein. As a result, the altered protein cannot bind effectively to DNA, which disrupts the control of genes involved in myelin formation and maintenance. This disruption results in the loss of myelin (demyelination) and impaired transmission of nerve impulses. As a result, the functioning of peripheral nerves that connect the brain and spinal cord to muscles and to sensory cells that detect sensations such as touch, pain, heat, and sound is reduced, causing the signs and symptoms of Charcot-Marie-Tooth disease.

A particular mutation has been identified in individuals with type 1D Charcot-Marie-Tooth disease who also have hearing loss. As a result of this mutation, the amino acid arginine is replaced with the amino acid histidine at protein position 381 (written as Arg381His). Other *EGR2* gene mutations cause a severe form of type 1D (sometimes called Dejerine-Sottas syndrome) that begins during infancy or early childhood. One of these mutations replaces the amino acid arginine with the amino acid tryptophan at protein position 359 (written as Arg359Trp). Another replaces the amino acid aspartic acid with the amino acid tyrosine at protein position 383 (Asp383Tyr). It is unclear why these particular mutations cause severe symptoms that begin early in life.

Chromosomal Location

Cytogenetic Location: 10q21.3, which is the long (q) arm of chromosome 10 at position 21.3

Molecular Location: base pairs 62,811,996 to 62,819,167 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- early growth response 2 (Krox-20 homolog, Drosophila)
- EGR2_HUMAN
- KROX20

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Early response genes function as development-control signals
  https://www.ncbi.nlm.nih.gov/books/NBK28157/#A1871

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28EGR2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- EARLY GROWTH RESPONSE 2
  http://omim.org/entry/129010

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EGR2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EGR2%5Bgene%5D
- HGNC Gene Family: Zinc fingers C2H2-type
  http://www.genenames.org/cgi-bin/genefamilies/set/28
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3239
- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=4
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1959
- UniProt
  http://www.uniprot.org/uniprot/P11161

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